Werner syndrome

Werner syndrome is characterized by the dramatic, rapid appearance of features associated with normal aging. Individuals with this disorder typically grow and develop normally until they reach puberty. Affected teenagers usually do not have a growth spurt, resulting in short stature. The characteristic aged appearance of individuals with Werner syndrome typically begins to develop when they are in their twenties and includes graying and loss of hair; a hoarse voice; and thin, hardened skin. They may also have a facial appearance described as "bird-like." Many people with Werner syndrome have thin arms and legs and a thick trunk due to abnormal fat deposition.

As Werner syndrome progresses, affected individuals may develop disorders of aging early in life, such as cloudy lenses (cataracts) in both eyes, skin ulcers, type 2 diabetes, diminished fertility, severe hardening of the arteries (atherosclerosis), thinning of the bones (osteoporosis), and some types of cancer. It is not uncommon for affected individuals to develop multiple, rare cancers during their lifetime. People with Werner syndrome usually live into their late forties or early fifties. The most common causes of death are cancer and atherosclerosis.

Frequency

Werner syndrome is estimated to affect 1 in 200,000 individuals in the United States. This syndrome occurs more often in Japan, affecting 1 in 20,000 to 1 in 40,000 people.

Genetic Changes

Mutations in the *WRN* gene cause Werner syndrome. The *WRN* gene provides instructions for producing the Werner protein, which is thought to perform several tasks related to the maintenance and repair of DNA. This protein also assists in the process of copying (replicating) DNA in preparation for cell division. Mutations in the *WRN* gene often lead to the production of an abnormally short, nonfunctional Werner protein. Research suggests that this shortened protein is not transported to the cell's nucleus, where it normally interacts with DNA. Evidence also suggests that the altered protein is broken down more quickly in the cell than the normal Werner protein. Researchers do not fully understand how *WRN* mutations cause the signs and symptoms of Werner syndrome. Cells with an altered Werner protein may divide more slowly or stop dividing earlier than normal, causing growth problems. Also, the altered protein may allow DNA damage to accumulate, which could impair normal cell activities and cause the health problems associated with this condition.

Inheritance Pattern

Werner syndrome is inherited in an autosomal recessive pattern, which means both copies of the *WRN* gene in each cell have mutations. The parents of an individual with Werner syndrome each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Adult premature aging syndrome
- Adult Progeria
- Werner's Syndrome
- Werners Syndrome
- WS

Diagnosis & Management

These resources address the diagnosis or management of Werner syndrome:

- GeneReview: Werner Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1514
- Genetic Testing Registry: Werner syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0043119/

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Health Topic: Cancer
  https://medlineplus.gov/cancer.html
- Health Topic: Cataract
  https://medlineplus.gov/cataract.html
- Health Topic: Coronary Artery Disease
  https://medlineplus.gov/coronaryarterydisease.html
- Health Topic: Osteoporosis
  https://medlineplus.gov/osteoporosis.html

Genetic and Rare Diseases Information Center

- Werner's syndrome
  https://rarediseases.info.nih.gov/diseases/7885/werners-syndrome

Additional NIH Resources

- National Heart, Lung, and Blood Institute: Atherosclerosis
  https://www.nhlbi.nih.gov/health/health-topics/topics/atherosclerosis/

Educational Resources

- Disease InfoSearch: Werner's syndrome
  http://www.diseaseinfosearch.org/Werner%27s+syndrome/7471
- International Registry of Werner Syndrome
  http://www.wernersyndrome.org/registry/registry.html
- MalaCards: atypical werner syndrome
  http://www.malacards.org/card/atypical_werner_syndrome
- MalaCards: werner syndrome
  http://www.malacards.org/card/werner_syndrome
- Merck Manual Consumer Version: Disorders of Accelerated Aging
  http://www.merckmanuals.com/home/older-people-s-health-issues/the-aging-body/disorders-of-accelerated-aging
- Orphanet: Werner syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=902
- University of Washington Werner Syndrome Research Website
  http://www.wernersyndrome.org/

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/werner-syndrome/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/werner.html

GeneReviews

- Werner Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1514

Genetic Testing Registry

- Werner syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0043119/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22werner+syndrome%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Werner+Syndrome%5BMAJR%5D%29+AND+%28Werner+syndrome%5BTIAB%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- WERNER SYNDROME
  http://omim.org/entry/277700

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